



GJA1 gene

gap junction protein alpha 1

Normal Function

The *GJA1* gene provides instructions for making a protein called connexin43, which is one of 21 connexin proteins in humans. Connexins play a role in cell-to-cell communication by forming channels, or gap junctions, between cells. Gap junctions allow for the transport of nutrients, charged particles (ions), and other small molecules that carry necessary communication signals between cells. Connexin43 is found in many human tissues such as the eyes, skin, bone, ears, heart, and brain.

Health Conditions Related to Genetic Changes

[critical congenital heart disease](#)

[heterotaxy syndrome](#)

[oculodentodigital dysplasia](#)

More than 45 mutations in the *GJA1* gene have been found to cause oculodentodigital dysplasia. Most of these mutations change one protein building block (amino acid) in connexin 43. A different change in the *GJA1* gene causes people to have oculodentodigital dysplasia with palmoplantar keratoderma. Palmoplantar keratoderma is a condition that causes skin on the palms and the soles of the feet to become thick, scaly, and calloused. This mutation deletes two DNA building blocks (nucleotides) to create a premature stop signal in the instructions for making connexin43. As a result, an abnormally short, nonfunctional protein is produced.

Channels formed with abnormal connexin 43 proteins are often permanently closed, preventing the transport of any molecules. Some mutations prevent connexin 43 proteins from traveling to the cell surface where they are needed to form channels. These disruptions in channel function impair communication between cells, which is thought to cause the eye, teeth, and finger abnormalities characteristic of oculodentodigital dysplasia.

[other disorders](#)

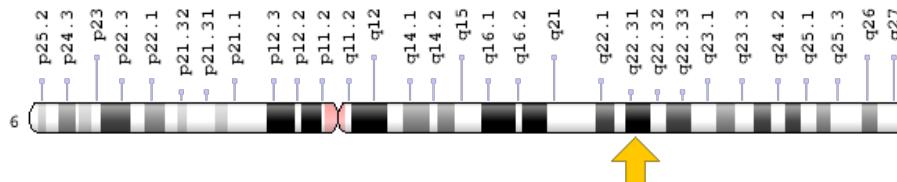
GJA1 gene mutations have been found to cause deafness in a small number of African Americans. Further research is needed to determine how common *GJA1*

mutations are among African-Americans and people in other ethnic groups with deafness.

Chromosomal Location

Cytogenetic Location: 6q22.31, which is the long (q) arm of chromosome 6 at position 22.31

Molecular Location: base pairs 121,435,577 to 121,449,744 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- connexin 43
- connexin43
- CX43
- Cx43 α 1
- CXA1_HUMAN
- gap junction 43 kDa heart protein
- gap junction protein, alpha 1, 43kDa
- gap junction protein, alpha-like

Additional Information & Resources

Educational Resources

- Madam Curie Bioscience Database: Connexins
<https://www.ncbi.nlm.nih.gov/books/NBK6455/>
- Molecular Biology of the Cell (fourth edition, 2002): Gap junctions
<https://www.ncbi.nlm.nih.gov/books/NBK26857/?rendertype=figure&id=A3497>

- The University of Western Ontario: Laird Lab
http://www.uwo.ca/anatomy/laird/research/connexin_mutations_and_disease.htm
- Washington University, St. Louis: Neuromuscular Disease Center
<http://neuromuscular.wustl.edu/spinal/fsp.html#oddd>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GJA1%5BTIAB%5D%29+OR+%28connexin+43%5BTI%5D%29+OR+%28CX43%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- GAP JUNCTION PROTEIN, ALPHA-1
<http://omim.org/entry/121014>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_GJA1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=GJA1%5Bgene%5D>
- HGNC Gene Family: Gap junction proteins
<http://www.genenames.org/cgi-bin/genefamilies/set/314>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4274
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2697>
- UniProt
<http://www.uniprot.org/uniprot/P17302>

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